Choices in Prenatal Testing for Women 35 Years and Older

There is a different booklet for ***women under 35*** years old. These women should ask their doctor or clinic for the blue and white booklet called: **"The California Expanded AFP Screening Program."**

Choices

California Department of Health Services—Genetic Disease Branch

The California Expanded AFP Screening Program is voluntary.
Women can refuse testing without losing eligibility
or services from State programs.

California Department of Health Services
Genetic Disease Branch
2151 Berkeley Way, Annex 4
Berkeley, CA 94704
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Choices in Prenatal Testing for Women 35 Years and Older

This booklet has information which will help pregnant women 35 years and older (at delivery) choose between:

- **diagnostic tests at a State-approved Prenatal Diagnosis Center**

or

- **the Expanded AFP Screening Program which includes a blood test— and diagnostic tests if the blood test is positive.**

After reading this pamphlet, talk with a genetic counselor or your doctor about what is best for you.

Every pregnant woman wonders about the health of her fetus (unborn baby) and the possibility of birth defects. Women 35 and older may be especially concerned because certain birth defects (such as Down syndrome) are more common in the pregnancies of older women.

The four **prenatal tests** described in this booklet can detect **some** birth defects but **not all of them**. Down syndrome, open neural tube defects, abdominal wall defects, trisomy 18 and other chromosomal defects are some of the birth defects found during testing. (These birth defects are described on pages 10 and 11.)

First, a woman needs to decide if she wants a screening test or a diagnostic test. A **diagnostic test** can tell whether or not the fetus **actually has** a certain birth defect. A **screening test** estimates the **chances (risk)** of the fetus having a certain birth defect. If the risk is high, a woman can then choose to have a diagnostic test.

The different tests are:

Diagnostic Tests	Screening Test
Amniocentesis Early Amniocentesis Chorionic Villus Sampling (CVS)	Expanded AFP Blood Test

Who can help you make this decision?

Before deciding between a screening test and a diagnostic test, a woman who will be 35 years or older at delivery should talk to her doctor and/or genetic counselor. A genetic counselor can explain your choices in detail and answer any questions. Your doctor or clinic can refer you to a genetic counselor at a State-approved Prenatal Diagnosis Center.

Who else besides women age 35 and older should have genetic counseling?

- women with a medical or family history of inherited conditions
- women who know that the baby's father has a medical or family history of inherited conditions
- women who are taking certain medicines
- women who have insulin-dependent diabetes

Payment for Services

Some health plans (including Medi-Cal) may only cover certain prenatal tests and counseling services. Get information from your insurance company or health plan about your coverage. (See page 12.)

Description of Diagnostic Tests

Amniocentesis

This test involves removing a small amount of the amniotic fluid that surrounds the fetus. It is usually done **between 15 and 20 weeks** of pregnancy. First, an ultrasound picture locates the fetus and the fluid. Then a thin needle is inserted through the woman's abdomen to remove a small amount of the fluid from the uterus.

In the fluid are cells from the fetus. The chromosomes in these cells are examined for Down syndrome and other chromosomal defects. Also, the fluid can be tested for neural tube defects. (See pages 10-11.) There is a small risk of miscarriage after amniocentesis: generally less than 1 in 100.

Early amniocentesis is done between **13 and 14 weeks** of pregnancy. There may be a slightly higher risk of miscarriage.

The results for these tests are available in about two weeks. Amniocentesis is over 99% accurate in diagnosing Down syndrome and other chromosomal defects. Amniocentesis detects nearly all open neural tube defects.

The cost of these tests is about $1,000 to $1,500. The patient or her insurance is responsible for payment. The Expanded AFP Screening Program does not pay for amniocentesis unless authorized. (See page 8.)

CVS (Chorionic Villus Sampling)

This test involves obtaining cells from the developing placenta instead of from the amniotic fluid. It is done early in pregnancy, **between 10 and 12 weeks**. An ultrasound picture locates the placenta in the uterus. A tiny piece of tissue is then removed from the placenta. This is done using a thin needle through the abdomen or a slender tube through the cervix. The cells from the placenta are examined for chromosomal defects.

There is a small risk of miscarriage: about 1 to 3 per 100. The final results are available in about two weeks. CVS is 98% accurate for the diagnosis of Down syndrome and other chromosomal defects. CVS cannot detect neural tube defects. An AFP blood test is recommended at 15-20 weeks of pregnancy to screen for these defects.

The cost for CVS is about $1,200 to $1,800. The patient or her insurance is responsible for payment. The Expanded AFP Screening Program does not pay for CVS.

Description of the Screening Test

The California Expanded AFP Screening Program

For many years, the only prenatal test for birth defects in women 35 years and older was amniocentesis. More recently, early amniocentesis and CVS have also been offered. There is another choice, as well: the **Expanded AFP blood test**.

This blood test result is combined with a woman's age to calculate her own personal risk for having a fetus with Down syndrome or trisomy 18 in this pregnancy. **Knowing this risk can help a woman decide whether to have amniocentesis.** The blood test result also provides information about the risk of open neural tube defects and abdominal wall defects.

Each pregnancy has its own risk, so results from previous tests do not apply to this pregnancy.

If the result is "screen positive," diagnostic follow-up tests are provided at no additional cost. (See page 8.)

If the Expanded AFP blood test is **"screen negative,"** the Program does **not** pay for any follow-up diagnostic tests.

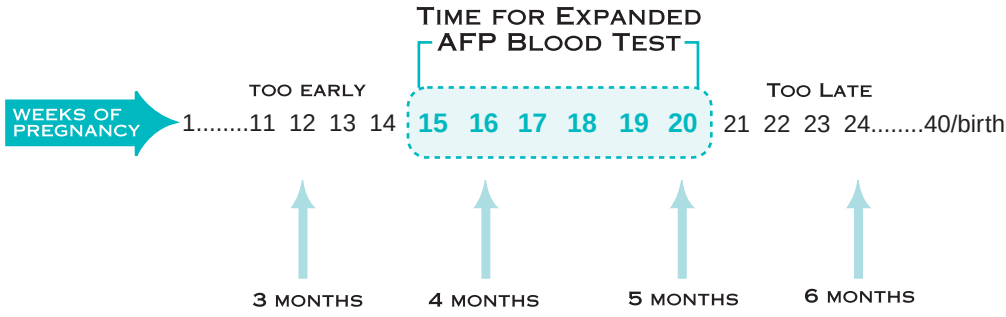
What does the blood screening test involve?

A small amount of blood is taken from the pregnant woman's arm. Her blood is tested for the amount of **AFP** (alpha-fetoprotein), **HCG** (human chorionic gonadotropin), and **UE** (unconjugated estriol). These substances are made by the mother's placenta and the fetus. At each week of pregnancy there are different amounts of these substances in the mother's blood. (What she eats does not affect these substances.)

When is the blood screening test done?

The blood test can only be done reliably **between 15 and 20 weeks of pregnancy**. *The best time is 16 to 17 weeks.* It is important to know how far along the pregnancy is. Ultrasound is very useful for this purpose.

The result of the blood test is sent to the patient's doctor or clinic within 1-2 weeks.



What does a "screen negative" result mean?

It means that the risk for *certain* birth defects is low enough that the Program does not consider follow-up tests necessary. The risk is calculated by measuring the amounts of AFP, HCG and UE in the woman's blood and also by considering her age.

Since the blood test is just a screening test, *there is still a chance that the fetus may have a problem* — even when the test result is "screen negative."

A "screen negative" result is the most common result.

About 9 out of 10 women tested will have a "screen negative" result.

- Because the risk for Down syndrome varies with a woman's age, so does the chance of a "screen negative" result. For example, among 35 year old women, about 9 out of 10 will have a "screen negative" result. Among 40 year old women, about 6 out of 10 will have a "screen negative" result.
- About 98 out of 100 women of all ages will have a "screen negative" result for neural tube defects and abdominal wall defects.
- About 99 out of 100 women of all ages will have a "screen negative" result for trisomy 18.

(See pages 10-11 for information about these birth defects.)

What does a "screen positive" result mean?

It means that the risk for certain birth defects in this pregnancy is higher than usual (including Down syndrome, neural tube defects, abdominal wall defects, and trisomy 18). The risk is calculated using the amounts of AFP, HCG and UE found in the woman's blood. Her age is part of the calculation for the risk of Down syndrome and trisomy 18.

Most of the time, however, the reason for the result is ***not*** a birth defect. The **most common reasons** for a "screen positive" result include:

- the due date is earlier or later than thought, *or*
- there is more than one fetus (twins, triplets), *or*
- the substances in the blood varied more than usual, without any known pregnancy problem.

To determine the reason for the "screen positive" result, **genetic counseling and follow-up diagnostic tests are offered** and paid for by the Program. Since receiving this result may cause anxiety, it is important to remember that *most women with "screen positive" results will have normal follow-up tests and healthy babies.*

Summary of Test Results

"screen negative" →	No follow-up tests are offered by the Program.
"screen positive" →	Follow-up tests are provided at no extra cost at a State-approved Prenatal Diagnosis Center.

If the test is "screen positive," what happens then?

A woman with a "screen positive" result will be **called by her doctor or clinic**. She will be offered diagnostic services at a **State-approved Prenatal Diagnosis Center**. When authorized, these are the follow-up services covered by the Program:

- **Genetic counseling** - A professional counselor discusses the pregnancy and family medical history. Questions are answered to help the woman make decisions about further testing.
- **Ultrasound** - A picture of the fetus is made using sound waves. This picture shows the age of the fetus and whether there are twins. The detailed ultrasound done at a Prenatal Diagnosis Center can also detect certain birth defects.
- **Amniocentesis** - A small amount of fluid is taken out of the uterus by experienced, State-approved doctors. The fluid and the fetal cells in it are tested for specific birth defects.

Women may refuse any of these services at any time.

What if the follow-up tests show that the fetus has a birth defect?

Information will be given to the woman by a doctor or genetic counselor at the Prenatal Diagnosis Center. They will discuss the type of birth defect that has been found and any available treatments. They will also discuss options for continuing or ending the pregnancy. The woman can then make a decision.

The Expanded AFP Screening Program does not pay for any other medical services after the follow-up tests. Referrals for special support services are available.

Birth Defects Found by the Program

What birth defects may be found through follow-up testing?

Down syndrome, open neural tube defects, abdominal wall defects, trisomy 18, and some other birth defects may be found.

Down Syndrome

Down syndrome is a common cause of mental retardation. Heart defects are often present, as well. Down syndrome can occur in the fetus of a woman of any age.

Down syndrome is caused by an extra chromosome #21. Chromosomes are packages of genetic material found in every cell of the body. Birth defects can occur when there are too few or too many chromosomes.

As women get older, their chances increase for carrying a fetus with Down syndrome.

For example, on average:

- 35 year old women have about 1 chance in 384 of having a child with Down syndrome.
- 40 year old women have about 1 chance in 112.

Not all cases of Down syndrome are found by the Program. In women 35 and older, 70% to 90% of the cases of Down syndrome are detected.

★ **Important Note:** The screening test is never 100% successful in detecting Down syndrome pregnancies. Only the diagnostic tests described on pages 3 and 4 will find almost 100% of them.

Birth Defects Found by the Program

Neural Tube Defects (NTDs)

As a fetus is forming, the neural tube extends from the top of the head to the end of the spine. This becomes the baby's brain and spinal cord. The neural tube is completely formed by 5 weeks after conception.

An opening in the spine is called **spina bifida**. This defect often causes paralysis of the legs. It may also cause loss of bowel and bladder control. Frequently, there is water-on-the-brain (hydrocephaly) which requires surgery.

Anencephaly occurs when most of the brain does not develop. This defect causes the death of the fetus or newborn.

Abdominal Wall Defects

Fetuses with these defects have **abnormal openings** on the abdomen. Intestines and other organs are formed outside the body. Surgery after birth often corrects the defect.

Trisomy 18

Trisomy 18 is caused by an extra chromosome #18. Babies with trisomy 18 have severe mental retardation and physical defects. They usually die before birth or in early infancy.

How Many Birth Defects Are Found?

These birth defects **do not occur very often**. However, **if there is one of these birth defects**, the Expanded AFP Screening Program helps detect it. Among women who have the Expanded AFP blood test and follow-up tests, the Program finds:

- 97% of the cases of anencephaly
- 80% of the cases of open spina bifida
- 85% of the cases of abdominal wall defects
- 60% of the cases of trisomy 18
- 70-90% of the cases of Down syndrome occurring in the pregnancies of women 35 years of age and older

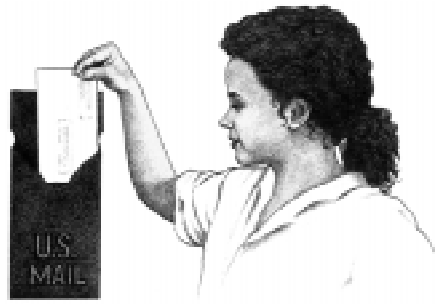
Can the Expanded AFP Screening Program detect every type of birth defect?

No. There are birth defects which cannot be detected by Expanded AFP Screening. Even when the blood test is "screen negative," there is still a chance the fetus may have a problem.

How much does the Expanded AFP Screening Program cost?

As of 1998, the fee is **$105**. (Check with the doctor or clinic about the most current fee.) The fee covers the blood test ***and*** authorized follow-up services at a State-approved Prenatal Diagnosis Center.

For women who already have had CVS (chorionic villus sampling) or early amniocentesis during this pregnancy, the current fee is $57. These women only have AFP testing. They do not need the HCG and UE tests.



The Program mails a bill to women who have the blood test. Women with private insurance should submit the bill to their insurance company. Women who have a prepaid health plan (HMO) may not receive a bill. If they do, they should send the bill to their health plan office. **As of January 1, 1999, insurance companies and HMOs are required to pay for Expanded AFP testing.**

Women with Medi-Cal usually do not receive a bill. If they do, they should return the bill with their Medi-Cal number. Women without insurance may make monthly payments and are responsible for the whole amount.

Any charges for drawing blood are <u>not</u> included in the program fee.

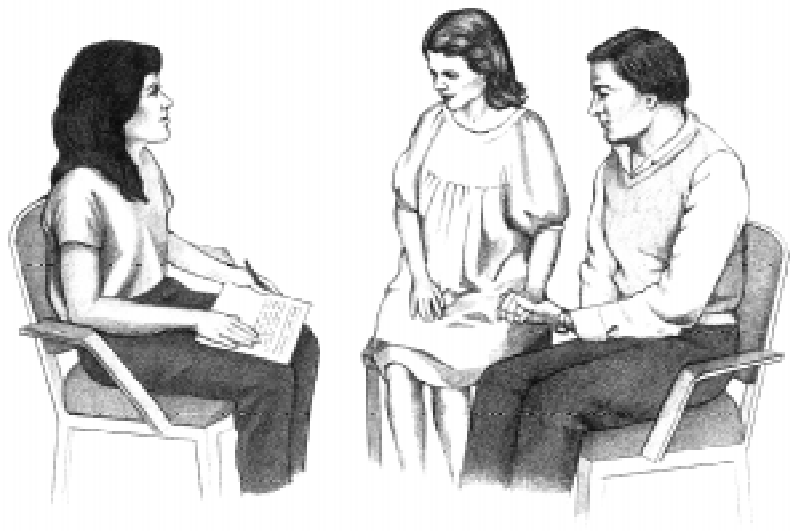
Summary of Prenatal Testing Choices

	Test	When Done	Description	Cost
Diagnostic Tests	CVS	10-12 weeks of pregnancy	• Diagnoses 98% of Down syndrome and other chromosomal defects • Not a test for neural tube defects, so an AFP blood test is recommended at 15-20 weeks • Chance of miscarriage is about 1%-3%	Approx. $1200 to $1800 plus $57 for AFP blood test
	Early Amniocentesis Amniocentesis	13-14 weeks of pregnancy 15-24 weeks of pregnancy	• Diagnoses 99% of Down syndrome and other chromosomal defects • Also diagnoses most open neural tube defects, abdominal wall defects, and some other birth defects • Chance of miscarriage is about 1%	Approx. $1000 to $1500
Screening Test	Expanded AFP Screening Program	15-20 weeks of pregnancy	• Estimates a woman's chance of having a fetus with Down syndrome or trisomy 18 • Also detects most open neural tube defects and abdominal wall defects • If the result is "screen positive," the Program pays for amniocentesis at a State-approved Prenatal Diagnosis Center. • Accurately predicts 70% to 90% of fetuses with Down syndrome in women 35 and older. Will miss detecting some Down syndrome and other chromosomal defects.	$105

Each woman should consider her prenatal testing choices carefully.

- Women who decide to have the Expanded AFP blood test must sign the consent form on the next page and have blood drawn between 15 and 20 weeks.
- Women who decide to have amniocentesis, CVS, or early amniocentesis should make an appointment at a State-approved Prenatal Diagnosis Center.
- Women should see a genetic counselor if they need help deciding between a screening test and diagnostic tests.
- Women can decide to have no prenatal testing.

Each woman should check with her insurance company or prepaid health plan about payment for these choices.



CLINICIAN'S COPY
(Remove and file in patient's chart.)

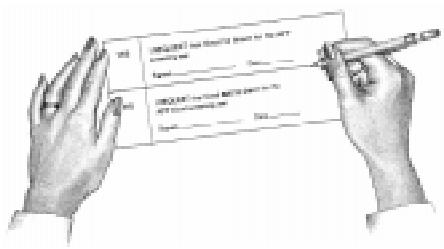
Patient's name_____
(PLEASE PRINT)

ID#_____

CONSENT/REFUSAL

FOR THE CALIFORNIA EXPANDED AFP SCREENING PROGRAM

1. I have read the information about the **California Expanded AFP Screening Program** which is contained in this booklet (or have had it read to me by_____).
2. I have been informed that:
   a) the purpose of the California Expanded AFP Screening Program is to detect most fetuses with Down syndrome, open neural tube defects, abdominal wall defects, and trisomy 18. However, not all such defects can be detected by the Program.
   b) there are other birth defects that cannot be detected by this Program.
   c) if the result is "screen positive," I will need to make a decision regarding follow-up testing. Authorized follow-up tests are covered by the Program and will be discussed with me in more detail.
   d) if the result is "screen negative," the Program will not pay for any follow-up testing.
   e) if the fetus is found to have a birth defect, the decision to continue or terminate the pregnancy will be entirely mine.
   f) participation in the California Expanded AFP Screening Program is voluntary. I can refuse any tests at any time.



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3. I have read the detection rates for certain birth defects as they are described in this booklet.
4 I have been informed that a blood specimen for the California Expanded AFP Screening Program is only reliable between 15 and 20 weeks of pregnancy.
5. I have been informed about the option of having CVS or amniocentesis instead of the Expanded AFP blood test.
6. I have had my questions answered to my satisfaction.

YES	I request that blood be drawn for the Expanded AFP Screening Program. Signed_____ Date_____ I should have my blood drawn between _____ and _____ month day year month day year
NO	I request that blood **not** be drawn for the Expanded AFP Screening Program. Instead, I have chosen _____ Signed_____ Date_____

I understand that the blood specimen and information obtained during the testing process become the property of the California Department of Health Services. They may be used for program evaluation or research by the Department or Department-approved scientific researchers without identifying the person or persons from whom these results were obtained, unless I specifically prohibit such use in writing. All information procured by the Department of Health Services, or by any other person, agency or organization acting jointly with the Department in connection with such special studies, shall be confidential. I may obtain additional information about the study or prohibit the use of my specimen by writing George Cunningham, MD, MPH, Genetic Disease Branch, 2151 Berkeley Way, Annex 4, Berkeley, CA 94704.

If new information becomes available about a birth defect detected during this pregnancy, the information may be sent to me unless I specifically prohibit it by writing to George Cunningham, MD, MPH at the above address.

TEAR HERE AND FILE IN PATIENT'S CHART

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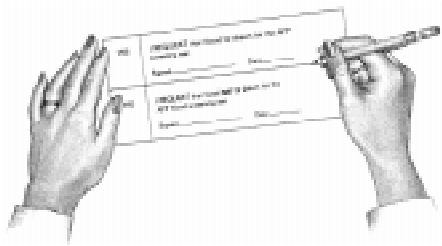

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Please keep this booklet and your copy of the consent/refusal form for your records.

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April 2000